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CONFERENCE ABSTRACT

The transition of patients with rare diseases between providers: the patient journey from the patient perspective.

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Background: Rare diseases are a group of more than 6000 disorders that on the whole may affect 30 million European Union citizens. Most rare diseases are of genetic origin, are often chronic and life-threatening. Patients living with rare diseases typically receive care from many providers and move frequently within health care settings, so high-quality transitional care is especially important for them, as well as for their family caregivers. Poor communications, incomplete transfer of information, inadequate education of health professionals, limited access to expertise services, and the absence of a single point person to ensure continuity of care all contribute to gaps in care during transitions.

The research project focuses on four rare Lysosomal storage disease - LSDs (Pompe disease, Anderson-Fabry disease, Gaucher disease and Mucopolysaccharidosis type 1) featured by the availability of treatments that allow for a good quality of life for patients and aims to gather information on the real patient journey and on the health and social services used in order to analyse and identify the key conditions for improving the care management of rare and ultra-rare diseases (RD).

Methods: We conducted a national survey, distributed to patients and caregivers from the 1st of June 2016 to the 7th of August 2016. Data were gathered through questionnaires disseminated online (through the patient associations' websites, Facebook pages and other online networks) or administered directly to patients through the patients' associations, to maintain the privacy and anonymity. The questionnaire consisted of 70 questions related to history and pattern of referrals to specialist, time to diagnosis, core medical tests, disciplines and specialists, time and type of treatment. The questionnaire was developed based on 16 in-depth interviews to patients sampled to cover the 4 LSDs, different ages and residence at the national level and further tested through the support of patients' associations. Data were analysed by descriptive and analytical statistics.

Results: Of the survey participants, 177 patients provided evaluable data. The sample covered the national territory, was mainly composed of adult patients (average 40 age), who were diagnosed with rare LSDs 13 years ago. According to the survey patients living with rare LSD diseases visited an average of 2.4 centres before receiving an accurate diagnosis and the mean length of time from symptom onset to accurate diagnosis was around 7.3 years, however, there

was a significant relationship between mean length of time for diagnosis and age (P >0.001), the mean length of time was reduced to 1.5 years for young patients (under 20 age), while was around 12 years for the older ones (over 65 age), indicating therefore the clear improvement in the diagnostic phase and in the access to reference centres. The analysis revealed the variability in tackling rarity and complexity, the wide number and specialization of professionals involved and the difficulty to provide integrated care pathways (ICPs) due to the lack of scalability and standardisation of care processes.

Conclusion: The combination of rarity, complexity and lack of effective treatment creates huge obstacles to the provision of holistic care and in many cases significant medical, psychological and social needs remain unmet. Rare and ultra-rare disease challenge the most traditional care management models, indeed, people with a RD often need follow up care and support from different categories of health professionals, often from several different medical specialities, as well as by social workers and other social and local service providers which requires a level of coordination not easy to organise in most health care systems.

Keywords: lysosomal storage diseases; patient journey; management; rare disease; patient survey